

Scleroderma

Fast Facts

- Scleroderma differs from person to person but can be very serious.
- There are medications, as well as steps individuals can take, to ease the symptoms of Raynaud's phenomenon, skin problems and heartburn.
- Effective treatments are available for those with severe disease, including acute kidney disease, pulmonary hypertension, lung inflammation and gastrointestinal problems.
- It is important to recognize and treat organ involvement early on to prevent irreversible damage.
- Patients should see physicians with specialized expertise in the care of this complex disease.



Scleroderma is an autoimmune rheumatic disease affecting the skin and other organs of the body, meaning that the body's immune system is acting abnormally. The main finding in scleroderma is thickening and tightening of the skin and inflammation and scarring of many body parts, leading to problems in the lungs, kidneys, heart, intestinal system and other areas. There is still no cure for scleroderma but effective treatments for some forms of the disease are available.

Scleroderma is relatively rare. About 75,000 to 100,000 people in the U.S. have this disease; most are women between the ages of 30 and 50. Twins and family members of those with scleroderma or other autoimmune connective tissue diseases, such as lupus, may have a slightly higher risk of getting scleroderma. Children can also develop scleroderma, but the disease is different in children than in adults.

Although the underlying cause is unknown, promising research is shedding light on the relationship between the immune system and scleroderma. A great deal of research is also underway to find better treatments for scleroderma and, hopefully, someday a cure.

What is scleroderma?

Scleroderma (also known as systemic sclerosis) is a chronic disease that causes the skin to become thick and hard, a buildup of scar tissue, and damage to internal organs such as the heart and blood vessels, lungs, stomach and kidneys. The effects of scleroderma vary widely and range from minor to life-threatening, depending on how widespread the disease is and which parts of the body are affected.

The two main types of scleroderma are:

- **Localized scleroderma**, which usually affects only the skin, although it can spread to the muscles, joints and bones. It does not affect other organs. Symptoms include discolored patches on the skin (a condition called morphea); or streaks or bands of thick, hard skin on the arms and legs (called linear scleroderma). When linear scleroderma occurs on the face and forehead, it is called en coup de sabre.
- **Systemic scleroderma**, which is the most serious form of the disease, affects the skin, muscles, joints, blood vessels, lungs, kidneys, heart and other organs.

The cause of scleroderma is not known. Genetic factors (different genes) appear to be important in the disease. Although exposure to certain chemicals may play a role in some people having scleroderma, the vast majority of patients with scleroderma do not have a history of exposure to any suspicious toxins. The cause of scleroderma is likely quite complicated.

How is scleroderma diagnosed?

Diagnosis can be tricky because symptoms may be similar to those of other diseases. There is no one blood test or X-ray that can say for sure that you have scleroderma. To make a diagnosis, a doctor will ask about the patient's medical history, do a physical exam and possibly order lab tests and X-rays. Careful clinical evaluation is the primary method for monitoring scleroderma. X-rays and computerized tomography (CT) scans are used to look at bone abnormalities. Thermography can detect differences in skin temperature between the lesion and normal tissue. Ultrasound and magnetic resonance imaging (MRI) can aid soft tissue assessment. Some symptoms he or she will look for include:

- **Raynaud's phenomenon:** This term refers to color changes (blue, white and red) that occur in fingers (and sometimes toes), often after exposure to cold temperatures. It occurs when blood flow to the hands and fingers is temporarily reduced. This is one of the earliest signs of the disease; more than 90 percent of patients with scleroderma have Raynaud's. Raynaud's can lead to finger swelling, color changes, numbness, pain, skin ulcers and gangrene on the fingers and toes. People with other diseases can also have Raynaud's and some people with Raynaud's do not have any other disease.
- **Skin thickening, swelling and tightening:** This is the problem that leads to the name "scleroderma" ("Sclera" means hard and "derma" means skin). The skin may also become glossy or unusually dark or light in places. The disease can sometimes result in changes in personal appearance, especially in the face. When the skin becomes extremely tight, the function of the area affected can be reduced (for example, fingers).

- Enlarged red blood vessels on the hands, face and around nail beds (called “telangiectasias”)
 - Calcium deposits in the skin or other areas
 - High blood pressure from kidney problems
 - Heartburn; this is an extremely common problem in scleroderma
 - Other problems of the digestive tract such as difficulty swallowing food, bloating and constipation, or problems absorbing food leading to weight loss
 - Shortness of breath
 - Joint pain
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+ How is scleroderma treated?

While some treatments are effective in treating some aspects of this disease, there is no drug that has been clearly proven to stop, or reverse, the key symptom of skin thickening and hardening. Medications that have proven helpful in treating other autoimmune diseases, such as rheumatoid arthritis and lupus, usually don't work for people with scleroderma. Doctors aim to curb individual symptoms and prevent further complications with a combination of drugs and self-care. For example:

- Raynaud's phenomenon can be treated with drugs such as calcium channel blockers or drugs called PDE-5 inhibitors –sildenafil (Viagra®), tadalafil (Cialis®) –, which open up narrowed blood vessels and improve circulation. To prevent further damage, it's important to keep the whole body warm, especially fingers and toes. It's also important to protect fingertips and other skin areas from injury, which can happen even during normal daily activities.
- Heartburn (acid reflux) can be treated with antacid drugs, especially proton-pump inhibitors (omeprazole and others). These medications ease gastro-esophageal reflux disease (known as GERD).
- Scleroderma kidney disease can be treated with blood pressure medications called “angiotensin converting enzyme inhibitors” (ACE inhibitors). These can often effectively control kidney damage if started early and use of these drugs has been a major advance for treating scleroderma.
- Muscle pain and weakness can be treated with anti-inflammatory drugs such as glucocorticoids (prednisone), intravenous immunoglobulin (IVIg), and/or immunosuppressive medications. Physical and occupational therapy may be useful to maintain joint and skin flexibility. Early referral to therapy should be considered to help prevent the loss of joint motion and function.

There are two types of lung disease that patients with scleroderma may develop. The first type is called interstitial lung disease (scarring). There is evidence that cyclophosphamide is somewhat effective in treating the interstitial lung disease in scleroderma. Clinical trials are underway assessing the effectiveness of several other drugs for this problem.

The second type of lung disease seen in scleroderma is pulmonary arterial hypertension (high blood pressure in the arteries in the lungs). In the last 10 years, a number of drugs have become available to treat this condition, including prostacyclin-like drugs (epoprostenol, treprostinol, iloprost), the endothelin receptor antagonists (bosentan, ambrisentan), and PDE-5 inhibitors (sildenafil, vardenafil, tadalafil).

Much research is ongoing into new treatments for scleroderma. Patients and their families should know that experts remain optimistic and take comfort in the fact that work towards a cure will continue.

+ Broader health impact of scleroderma

Scleroderma can involve almost every organ system in the body. Although symptoms vary greatly from patient to patient, it can dramatically impact someone's life. Patients should consult a rheumatologist—or a team of specialists—who are experienced in dealing with this complicated disease. Several other diseases that affect the skin are sometimes confused with scleroderma.

+ Living with scleroderma

Living with scleroderma is quite challenging. Everyday activities can sometimes be difficult due to physical limitations and pain. Problems with digestion may require changes in diet; patients often have to eat several small meals rather than fewer large meals. Patients must also keep the skin well-moisturized to lessen stiffness and be careful during activities such as gardening, cooking—even opening envelopes—to avoid finger injuries. To keep the body warm, patients should dress in layers; wear socks, boots and gloves; and avoid very cold rooms. Unfortunately, moving to a warmer climate does not necessarily lead to dramatic improvement. Exercise and/or physical therapy may ease stiffness in the joints.

Patients must also deal with the psychological setbacks that come from living with a disease that is chronic, uncommon and currently incurable. Because scleroderma can cause significant changes in appearance, a patient's self-esteem and self-image are almost always affected. The support of family and friends is vital in helping to maintain a good quality of life.

Updated June 2015. Written by Peter A. Merkel, MD, MPH; and reviewed by the American College of Rheumatology Communications and Marketing Committee.

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